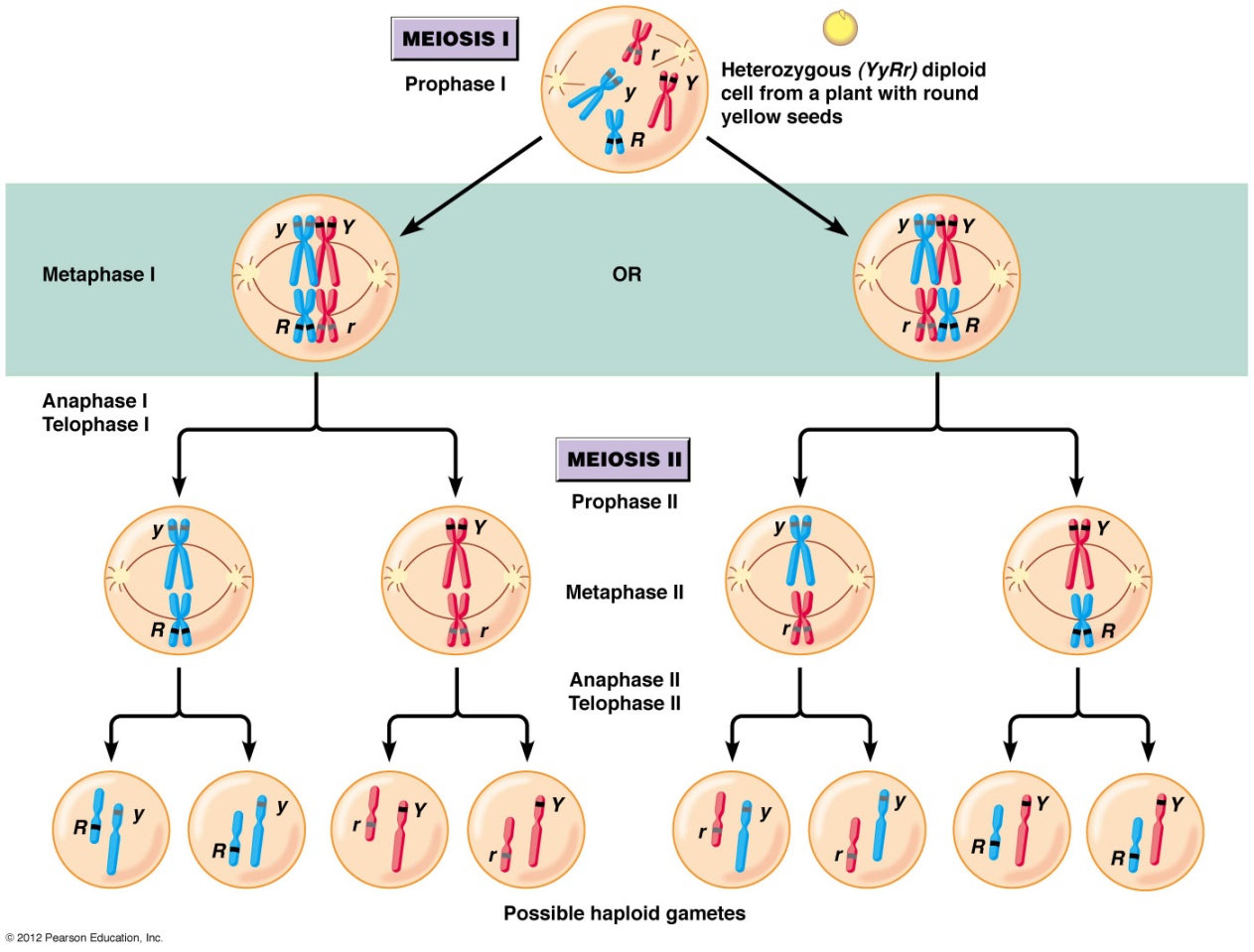
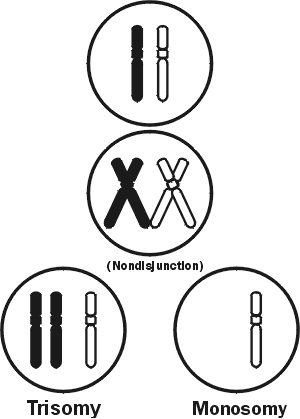
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**Unit 9, Part 3 Notes: Chromosomal Genetics**

AP Biology, Mrs. Krouse

1. **What are Mendel’s Laws of Inheritance?**
2. Gregor Mendel devised three laws of inheritance: The Law of Dominance, The Law of Segregation, and The Law of Independent Assortment.
3. The Law of Dominance states that a dominant allele completely masks the effects of a recessive allele. Therefore, when an individual has a heterozygous genotype (one dominant and one recessive allele), he/she shows the dominant phenotype only.
4. The Law of Segregation states that pairs of homologous chromosomes (and their alleles) are separated into gametes (eggs and sperm) during meiosis. Therefore, gametes only contain one allele for each gene. Below is an image showing meiosis and the inheritance of two genes, one for seed color (Y = yellow, y = green) and one for seed texture (R = round, r = wrinkled). Note that each of the gametes ends up with one copy of the seed color gene and one copy of the seed texture gene.
5. The Law of Independent Assortment states that genes on different pairs of homologous chromosomes separate independently into gametes. If genes are on different pairs of homologous chromosomes, the separation of one gene into a gamete is independent of the separation of the other gene into a gamete. This is because each pair of homologous chromosomes lines up independently of the others along the metaphase plate during Metaphase I of meiosis. The image below shows an individual with the following genotype RrYy. When determining the gametes that go on the top or side of a dihybrid Punnett square for this individual, we would use the FOIL method (see Unit 8 Notes, Part 1), giving us four possible gametes—RY, Ry, rY, and ry. The variety of possible gametes is a result of independent assortment. The pathway on the left in the image below shows one possible way the homologous chromosomes can line up, producing the gametes Ry and rY. The pathway on the right in the image below shows another possible way the homologous chromosomes can line up, producing the gametes ry and RY.

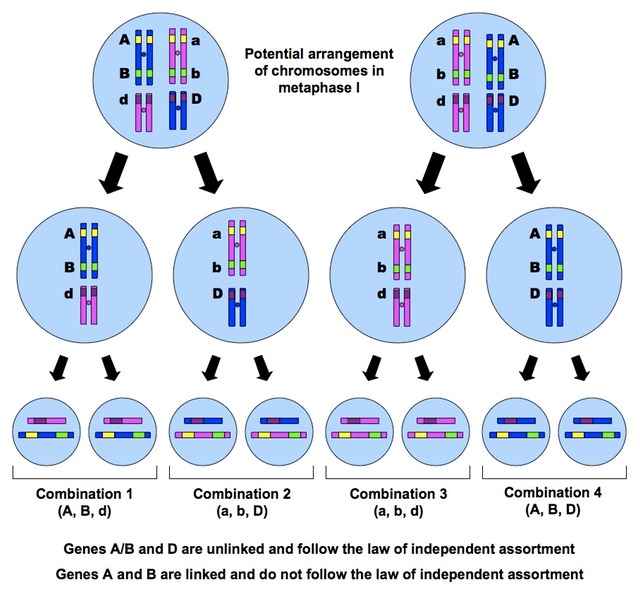


1. **What happens when alleles don’t segregate (separate) properly during meiosis?**
2. When chromosomes do not separate properly during meiosis, this is called nondisjunction. When this happens, gametes end up with an incorrect number of chromosomes, a condition called aneuploidy.
3. When a gamete ends up with both homologous chromosomes from a particular chromosome pair and this gamete is fertilized, the baby will have three copies of a particular homologous chromosome (one extra chromosome overall). This is called trisomy.

* When this happens to the 21st chromosome pair, the condition is called trisomy 21 (also known as Down Syndrome).
* When this happens to the sex chromosomes (the 23rd chromosome pair), an individual can end up having two X chromosomes and one Y chromosome (XXY). This condition is known as Klinefelter Syndrome.

1. When a gamete ends up with neither homologous chromosome from a particular chromosome pair and this gamete is fertilized, the baby will have only one copy of a particular homologous chromosome (one less chromosome overall). This is called monosomy.

* When this happens to the sex chromosomes (the 23rd chromosome pair), an individual can end up having only one X chromosome (XO). This condition is known as Turner Syndrome.

1. If nondisjunction occurs in ALL pairs of homologous chromosomes, this can result in a gamete with a full extra set of chromosomes. When this gamete is fertilized, it could result in a triploid baby (one with three sets of chromosomes instead of a normal baby that has two sets of chromosomes—i.e. a diploid organism).
2. Some species of plants end up with four sets of chromosomes (tetraploid) or more. When an organism as any number of extra sets of chromosomes, this is called polyploidy. For example, wheat plants have been hybridized and manipulated by humans, to the point where there are several strains—one that is diploid, one that is tetraploid, and one that is hexaploid. Polyploidy often results in speciation, the creation of species with different “ploidy’s” that can no longer mate. For some reason, polyploidy can result in species of plants that are more vigorous / healthy than the normal diploid species. Polyploidy has also been documented in fish and amphibians.
3. **How are genes inherited when they are located on the same chromosome?**
4. Genes that are located on the same chromosome do not follow the Law of Independent Assortment. Instead, they are always inherited together, unless they are separated by crossing over between homologous chromosomes. The image to the right, shows two genes (A and B) that are found on the same chromosome. These are called linked genes. Gene D is found on another chromosome, so it is considered “unlinked” to genes A and B.
5. Because they are found on the same homologous chromosome, alleles A and B always separate into gametes together. Because they are found on the same homologous chromosome, alleles a and B always separate into gametes together. For this individual with the genotype AaBb, this results in only two possible gamete combinations for these two genes—AB and ab. If the genes were unlinked, there would be four possible gamete combinations—AB, Ab, aB, and ab.
6. For this individual, there is a chance that the gametes Ab and aB can be produced if crossing over occurs between the pair of homologous chromosomes during prophase I of meiosis (i.e. the type of cell division that creates sex cells). Let’s say gene A controls height and that A = tall and a = short. Let’s say gene B controls eye color and that B = brown and b = blue. If two individuals with the genotype AaBb were crossed and the genes A and B were unlinked, the following phenotype ratio would result in the offspring.

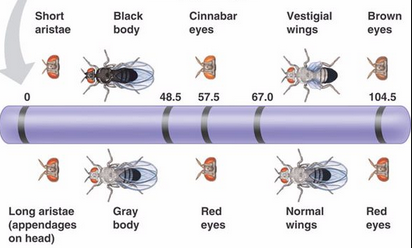
|  |  |
| --- | --- |
| **Phenotype** | **Number of Offspring** |
| Tall, Brown | 9 |
| Tall, Blue | 3 |
| Short, Brown | 3 |
| Short, Blue | 1 |

If two individuals with the genotype AaBb were crossed and the genes A and B are linked (as in the image on the previous page), the following phenotype ratio could result in the offspring (or a similar ratio).

|  |  |
| --- | --- |
| **Phenotype** | **Number of Offspring** |
| Tall, Brown | 7 |
| Tall, Blue | 1 |
| Short, Brown | 1 |
| Short, Blue | 7 |

The individuals that are tall / blue and short / brown resulting from separation of the linked genes by crossing over between homologous chromosomes.

1. The first linked genes were discovered by Thomas Hunt Morgan in fruit flies.
2. **How often are linked genes separated by crossing over?**
3. Genes that are located farther apart from one another on the same chromosome are more likely to be separated by crossing over.
4. Scientists can determine the frequency of genes being separated by crossing over by studying offspring phenotype ratios. The frequency of genes being separated by crossing over is known as the recombination frequency for those particular genes.
5. Recombination frequencies can be used to construct linkage maps showing the locations of gene loci (gene locations) along the length of a chromosome. The distance between two genes on a chromosome is measured in map units. 1 map unit corresponds to 1% recombination frequency. In other words, the genes are only separated by crossing over 1% of the time during meiosis.
6. Below is a linkage map of several genes located on a fruit fly chromosome.



1. Recombination frequencies below 50% correspond to genes located on the same chromosome, and recombination frequencies above 50% correspond to genes located on different chromosomes or genes that are so far apart on the same chromosome that their offspring phenotype ratios suggest they are unlinked.

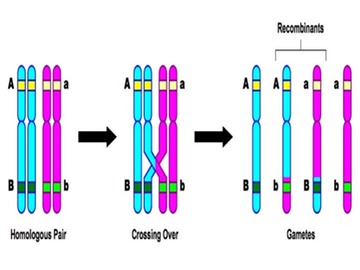
**Linked Genes Clarification**

**Definition of Linked Genes:**

When genes are linked, they are found on the same chromosome and are almost always inherited together, unless they are separated by crossing over (which is often called gene recombination). See image of two sets of linked alleles (A is linked to B, and “a” is linked to b) being separated by crossing over to create some “recombinants” (chromosomes with A and b, and chromosomes with “a” and B). Linked genes do not follow the rule of Independent Assortment because they are almost always inherited together. Therefore, when tracking the inheritance of unlinked genes, the offspring genotype and phenotype frequencies will NOT match the frequencies predicted by Punnett Squares. Typically the frequencies of individuals with “linked traits” are higher than the frequencies of individuals with “recombinant traits” because recombinant individuals can only be created through crossing over (see example and image below).

For example, if A codes for tall and B codes for brown hair, then tall and brown hair are linked traits (because A and B are on the same chromosome). If “a” codes for short and b codes for blond hair, then short and blond are also linked traits (because “a” and b are on the same chromosome). Therefore, tall and blond haired (A and b) individuals and short and brown haired individuals (“a” and B) can only be created through crossing over of the homologous chromosomes. Thus these “recombinant” individuals are less frequent than individuals who possess the linked gene combinations.

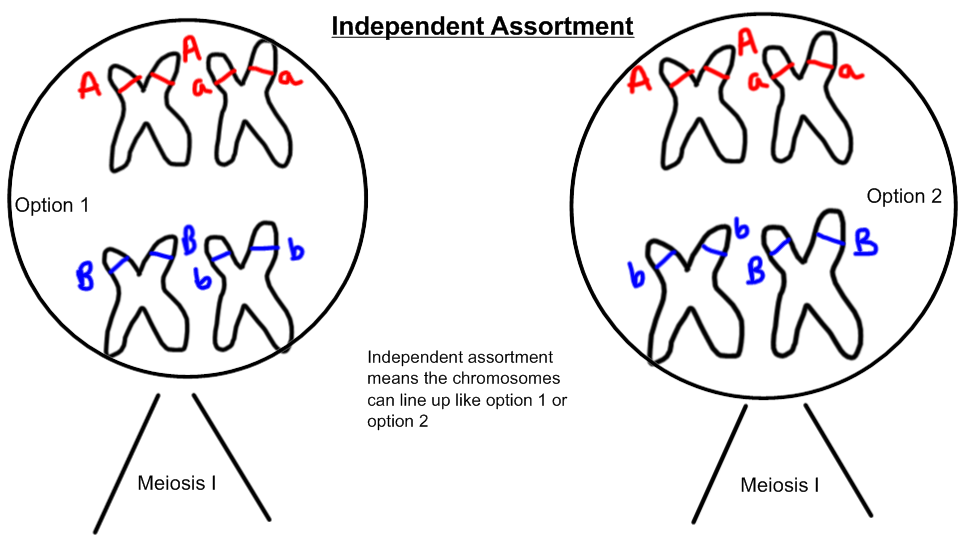
***Image #1:***

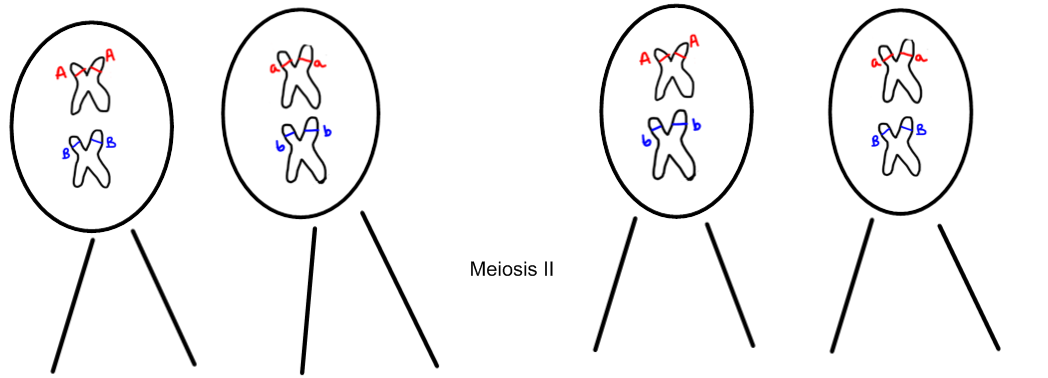


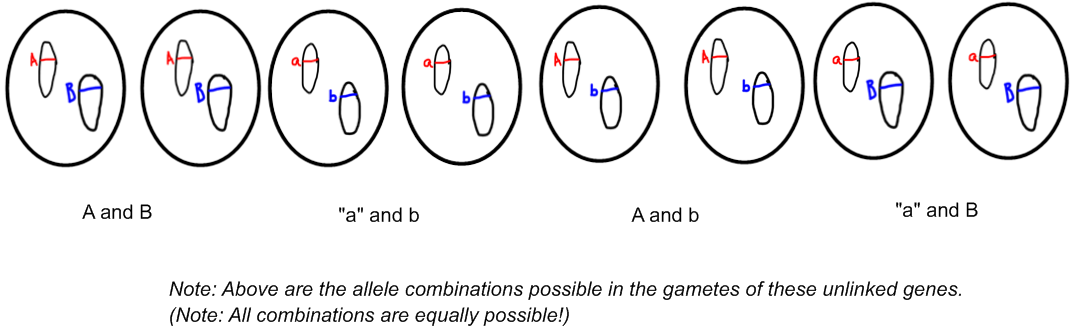
When genes are unlinked, they are found on different chromosomes. This means that they are not necessarily inherited together, and they follow the rule of independent assortment (each pair of homologous chromosomes is separated independently of the other chromosome pairs to enter the gametes). The image on the next page show independent assortment of unlinked genes A and B.

Note: As a result of independent assortment, A is just as likely to be inherited (i.e. put in the same gamete) with B as it is with b. Likewise, “a” is just as likely to be inherited with B as it is with b.

***Image #2:***







Therefore, when tracking the inheritance of unlinked genes, the offspring genotype and phenotype frequencies should match the frequencies predicted by Punnett Squares.

**Sample Genetics Problems Involving Linked vs. Unlinked Genes:**

Let’s say we are tracking the inheritance of ear length and fur color in rabbits. We will use “A” to represent the dominant long ear allele and “a” to represent the recessive short ear allele. We will use “B” to represent the dominant dark fur color allele and “b” to represent the recessive light fur color allele.

Let’s say we crossed two individuals with the genotypes AaBb and aabb. If the genes are not linked, we might see offspring phenotype frequencies like the ones given below…

***Table #1:***

|  |  |
| --- | --- |
| **Phenotypes** | **Number of Offspring** (out of 100) |
| Long Ears, Dark Fur | 23 (23%) |
| Long Ears, Light Fur | 28 (28%) |
| Short Ears, Dark Fur | 22 (22%) |
| Short Ears, Light Fur | 27 (27%) |

Note: These phenotype frequencies approximately match with the phenotype frequencies we would expect if we set up a dihybrid cross between the two parents.

***Table #2:***

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | AB | Ab | aB | ab |
| ab | *AaBb* | **Aabb** | aaBb | **aabb** |
| ab | *AaBb* | **Aabb** | aaBb | **aabb** |
| ab | *AaBb* | **Aabb** | aaBb | **aabb** |
| ab | *AaBb* | **Aabb** | aaBb | **aabb** |

I have color coded the offspring genotypes that will result in the four offspring phenotypes. A summary of the offspring phenotype ratios is given below.

Long Ears, Dark Fur (*italicized*) = 4/16 = 25%

Long Ears, Light Fur (**bolded**) = 4/16 = 25%

Short Ears, Dark Fur (underlined) = 4/16 = 25%

Short Ears, Light Fur (**bolded and underlined**) = 4/16 = 25%

You don’t actually need to use a dihybrid cross to solve this problem. You can use two monohybrid Punnett squares (one for the “A’s” and one for the “B’s”) and then multiply the frequencies of getting a particular ear phenotype with a particular fur phenotype to get the frequency of the two phenotypes happening together (i.e. the Multiplication Rule of Probabilities).

***Table #3:***

|  |  |  |
| --- | --- | --- |
|  | a | a |
| A | Aa | Aa |
| a | aa | aa |

***Table #4:***

|  |  |  |
| --- | --- | --- |
|  | b | b |
| B | Bb | Bb |
| b | bb | bb |

For example, the frequency of having long ears according to Table #3 is ½ and the frequency of having dark fur according to Table #4 is ½. To get the frequency of having both long ears and dark fur, you must multiply their individual probabilities.

½ x ½ = ¼ (or 25%)

If the offspring phenotype frequencies from Table #1 approximately match with the frequencies we would expect if we used Punnett squares, then the genes are unlinked (found on different chromosomes) and therefore DO follow the law of independent assortment, as shown in Image #2.

Let’s say we had different offspring phenotype frequencies for Table #1 (see below).

***Alternate Table #1***

|  |  |
| --- | --- |
| **Phenotypes** | **Number of Offspring** (out of 100) |
| Long Ears, Dark Fur | 45 (45%) |
| Long Ears, Light Fur | 6 (6%) |
| Short Ears, Dark Fur | 5 (5%) |
| Short Ears, Light Fur | 44 (44%) |

These frequencies do not match with the frequencies predicted by Punnett squares (25%, 25%, 25%, and 25%). Rather, two of the phenotypes (long ears / dark fur and short ears / light fur) are far more common than the other two phenotypes (long ears / light fur and short ears / dark fur). Therefore, the alleles for long ears and dark fur (A and B) are linked (found on the same chromosome). The alleles for short ears and light fur (“a” and b) are also linked. The other two possible phenotypes—long ears / light fur (A and b) and short ears / dark fur (“a” and B) are only created through crossing over, as shown in Image #1.

What if the parent genotypes are not AaBb x aabb? Let’s say our parent genotypes are AaBb and AaBb. If we completed a Punnett square analysis for these parent genotypes, we would have the following phenotype frequencies in our offspring: 9/16 (56%) long ears / dark fur, 3/16 (19%) long ears / light fur, 3/16 (19%) short ears / dark fur, and 1/16 (6%) short ears / light fur. If our frequencies from an actual experimental breeding are similar to the ones given below in Table #5, then the genes are unlinked, since they appear to match the frequencies predicted by Punnett squares.

If the genes are linked (A to B and “a” to b), we would expect to see frequencies like those given in Alternate Table #1, where two phenotypes (long ears / dark fur and short ears / light fur) are far more common than the other two phenotypes (long ears / light fur and short ears / dark fur).

***Table #5:***

|  |  |
| --- | --- |
| **Phenotypes** | **Number of Offspring** (out of 100) |
| Long Ears, Dark Fur | 58 (58%) |
| Long Ears, Light Fur | 18 (18%) |
| Short Ears, Dark Fur | 17 (17%) |
| Short Ears, Light Fur | 7 (7%) |

**Notes Questions**

Thank you to Ms. Glick

***For this notes packet, you must complete one and a half pages of notes annotations. This means you will complete the front and back of one page and the front of another.***

**Vocabulary:** The following terms have been chosen for you from the Part 3 Notes. Define each term in the set and identify a connection between the terms in the set.

1. Terms: Law of Dominance and Law of Segregation and Law of Independent Assortment

Definitions and Connection:

2.Terms: nondisjunction and aneuploidy

Definitions and Connection:

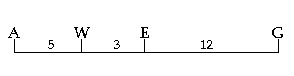
3.Terms: recombination frequency and linkage maps

Definitions and Connection:

**Ideas / Concepts:** The following are three main ideas/concepts from the Part 3 Notes. For each idea provide a one sentence explanation of that overarching idea.

1. *Consequences of improper separation of homologous chromosomes*
2. *The differences between linked and unlinked genes*
3. *How crossing over can be used to determine the location of genes on a chromosome*

**Supporting Evidence:** Choose one of the key ideas listed above and expand upon this main idea in 2-3 more sentences

1. Using the map to the right, between which two genes would you expect the lowest frequency of recombination?

****

1. Using the karyotype to the right, what types of information can you draw? (i.e. Does the person have a disorder? What type of disorder? What happened to produce this disorder? What else can you tell about this person?)

|  |  |
| --- | --- |
|  | F2 Generation |
| 165 | red eyes, straight wings |
| 168 | purple eyes, curly wings |
| 8 | purple eyes, straight wings |
| 6 | red eyes, curly wings |

1. A male fruit fly (*Drosophila melanogaster*) with red eyes and straight wings was mated with a female with purple eyes and curly wings. All of the offspring in the F1 generation had red eyes and straight wings. These F1 flies were test crossed with purple-eyed, curly-winged flies. Their offspring, the F2 generation, appeared as indicated to the right.
   1. Why is there a high frequency of red eyed /straight winged flies and purple eyed / curly winged flies?
   2. How is it possible to have purple eyed / straight winged flies and red eyed / curly winged flies?